# G6PD Deficiency

G6PD deficiency is a common genetic condition caused by defects in an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. The G6PD enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are destroyed upon exposure to certain environmental triggers, which can lead to episodes of anemia. This test includes the most common variant linked to G6PD deficiency in people of African descent.

## play+14746013e3, we could **not determine** if you have the genetic variant we tested.

This test is intended to detect one variant in the G6PD gene, but your result could not be determined.

This can be caused by random test error or other factors that interfere with the test. It can also happen when a person's genetic result is different from what is expected based on the sex they reported in their account settings. If this result is unexpected, please contact our Customer Care team.

## How To Use This Test

**This test does not diagnose G6PD deficiency or any other health conditions.**

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

### Intended Uses

- Tests for the **Val68Met** variant in the G6PD gene linked to G6PD deficiency.

### Limitations

- Does **not** test for all possible variants linked to G6PD deficiency.
- Your result for this report depends on the sex you indicated in your account settings.

### Important Ethnicities

- The variant included in this test is most common and best studied in people of **African** descent.
- This variant is also found in populations with African ancestry, like Hispanics or Latinos.
- This test does **not** include variants that are more common in people of Mediterranean, Middle Eastern, Asian, or Kurdish Jewish descent.

## We could **not determine** if you have the variant we tested linked to G6PD deficiency.

If this condition runs in your family, consider talking with a healthcare professional about additional testing.

### We could not rule out the variant we tested, called Val68Met.

This variant is most commonly found in people of **African** descent. It is also found in populations with African ancestry, like **Hispanics** or **Latinos**.

See Scientific Details

### You still have a chance of having this genetic variant or another variant linked to G6PD deficiency.

However, even people with variants linked to G6PD deficiency usually do not develop symptoms unless they are exposed to certain environmental triggers.

See Scientific Details

## For people with G6PD deficiency, symptoms can be triggered by certain factors.

### Certain medications

A number of medications can trigger symptoms in people with G6PD deficiency. For example, certain medications that are used to treat malaria, cancer, and bacterial infections can trigger symptoms in some people. People with G6PD deficiency should consult with a healthcare professional before starting or stopping any medications.

See Scientific Details for more information

## About G6PD Deficiency

**Also known as**: Glucose-6-Phosphate Dehydrogenase Deficiency, G6PDD

### When it develops

Because it is a genetic condition, G6PD deficiency is present at birth. However, people with this condition typically don't develop symptoms unless they are exposed to certain triggering factors. Many people with G6PD deficiency never develop symptoms.

### Typical signs and symptoms

- Anemia
- Dark urine
- Fatigue
- Pale skin
- Shortness of breath
- Jaundice (yellowing of the skin and eyes)

### How common is the condition?

Approximately 400 million people worldwide have G6PD deficiency. The condition occurs most frequently in people from certain parts of Africa, Asia, the Middle East, and the Mediterranean. About 1 in 10 African-American men has G6PD deficiency. Because of the way it is inherited, men are more likely to have this condition than women.

### How it's treated

Most people with G6PD deficiency do not require treatment. People with G6PD deficiency often manage their condition by avoiding certain medications and foods that may trigger symptoms. If a person is exposed to a trigger and develops anemia, symptoms usually clear up on their own. However, in some cases patients may require a blood transfusion.

**Read more at:** Genetic and Rare Diseases Information Center MedlinePlus Genetics Home Reference

## Consider talking to a healthcare professional if this condition runs in your family or you are concerned about your results.

See our Frequently Asked Questions for more information.

FAQs

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

Print report



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G6PD Deficiency

# Scientific Details

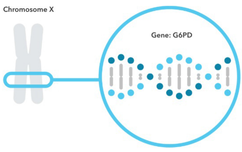
G6PD deficiency is a common genetic condition caused by defects in an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. The G6PD enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are destroyed upon exposure to certain environmental triggers, which can lead to episodes of anemia. This test includes the most common variant linked to G6PD deficiency in people of African descent.

## G6PD deficiency is linked to variants in the G6PD gene.

G6PD

The G6PD gene contains instructions for making an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. This enzyme is important for protecting red blood cells against damage. Certain variants in the G6PD gene reduce the levels or activity of the enzyme. This can lead to the destruction of red blood cells in response to certain medications, infections, and foods. The G6PD gene is located on the X chromosome, which means that males have one copy of this gene and females have two.

**Read more at** Genetics Home Reference*




## Your result for the genetic variant we tested could not be determined.

Variants Detected | View All Tested Markers

| Marker Tested | Your Genotype* | Additional Information |
|---|---|---|
| **Val68Met**<br>**Gene:** G6PD<br>**Marker:** rs1050828 | **Not determined** | Biological explanation<br>Typical vs. variant DNA sequence(s)<br>Percent of 23andMe customers with variant<br>**References** [ 4, 6, 8, 9, 11, 13, 17, 18, 19 ] \| ClinVar* |

*23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (Build 37). Other sources sometimes report genotypes using the opposite strand.

## Test Interpretation

This report provides information about risk for G6PD deficiency in people who have the variant included in this test.

### Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Consider talking to a healthcare professional if you have any concerns about your results.

**References** [ 4, 10, 15, 16 ]

**Risk estimates**

- The variant in this report is most commonly found in people of African descent. Depending on which part of Africa a person's ancestors are from, the chances of having a variant can be more than 20%.
- Not everyone with a variant has G6PD deficiency. Studies suggest that about 70% of males with the variant in this report have the condition, and about 50% of females with two copies of the variant have the condition. Females with just one copy of the variant are not typically at risk for developing symptoms of G6PD deficiency.
- People with G6PD deficiency usually don't have symptoms unless they are exposed to certain medicines, infections, or foods. Many people with the condition never experience symptoms.

## Other Factors

G6PD deficiency is a genetic condition. People with this condition are at risk for hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. Exposure to certain factors is usually required to trigger hemolytic anemia.

This is not a complete list of other factors.

Only people with certain variants in the G6PD gene are at risk for G6PD deficiency and its symptoms.

The factors described here include the most common and well-established risk factors associated with anemia in people with G6PD deficiency. Other factors not listed here may also influence risk for anemia in people with the condition. Specific triggers may vary from person to person, even among people with the same variant.

Consult with a healthcare professional before making any major lifestyle changes.

| Other Factors | References |
|---|---|
| **Certain medications**<br>A number of medications can trigger symptoms in people with G6PD deficiency. For example, certain medications that are used to treat malaria, cancer, and bacterial infections can trigger symptoms in some people. People with G6PD deficiency should consult with a healthcare professional before starting or stopping any medications. | [ 1, 2, 7, 14 ] |
| **Certain infections**<br>Certain infections can trigger symptoms in people with G6PD deficiency. These include bacterial infections, such as Salmonella and E. coli, as well as viral infections, such as hepatitis and flu. | [ 1, 2, 7, 14 ] |
| **Certain foods**<br>For some people with G6PD deficiency, eating certain foods such as fava beans (also called broad beans) can trigger symptoms. | [ 1, 2, 7, 14 ] |
| **Certain chemicals**<br>For some people with G6PD deficiency, being exposed to certain chemicals can trigger symptoms. These chemicals include naphthalene (which can be found in moth balls) and methylene blue (a dye used in some medical tests and scientific experiments). | [ 1, 2, 7, 12, 14 ] |

## Test Details

### Indications for Use

The 23andMe PGS Genetic Health Risk Report for G6PD Deficiency is indicated for reporting of the Val68Met variant in the G6PD gene. This report describes if a person has a variant associated with G6PD deficiency and a higher risk for episodes of anemia, but it does not describe a person's overall risk of developing symptoms. This report is most relevant for people of African descent.

**Special Considerations**

- This test does not include the Asn126Asp variant in the G6PD gene. The Asn126Asp variant is not linked to G6PD deficiency but is commonly included in genetic tests for this condition.
- Genetic testing for G6PD deficiency in adults in the general population is not currently recommended by any healthcare professional organizations.

### Test Performance Summary

**Clinical Performance** [ 3, 10 ]

The variant included in this test is expected to be responsible for up to 90% of cases of G6PD deficiency in people of African descent. This test does not include variants that are more common in people of Mediterranean, Middle Eastern, Asian, and Kurdish Jewish descent.

**Analytical Performance**

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

### Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

## References

1. Beutler E. (1994). "G6PD deficiency." Blood. 84(11):3613-36.*
2. Beutler E. (2008). "Glucose-6-phosphate dehydrogenase deficiency: a historical perspective." Blood. 111(1):16-24.*
3. Cappellini MD et al. (2008). "Glucose-6-phosphate dehydrogenase deficiency." Lancet. 371(9606):64-74.*
4. Carter N et al. (2011). "Frequency of glucose-6-phosphate dehydrogenase deficiency in malaria patients from six African countries enrolled in two randomized anti-malarial clinical trials." Malar J. 10:241.*
5. Clark TG et al. (2009). "Allelic heterogeneity of G6PD deficiency in West Africa and severe malaria susceptibility." Eur J Hum Genet. 17(8):1080-5.*
6. Dombrowski JG et al. (2017). "G6PD deficiency alleles in a malaria-endemic region in the Western Brazilian Amazon." Malar J. 16(1):253.*
7. Frank JE. (2005). "Diagnosis and management of G6PD deficiency." Am Fam Physician. 72(7):1277-82.*
8. Gomez-Gallego F et al. (2000). "Structural defects underlying protein dysfunction in human glucose-6-phosphate dehydrogenase A(-) deficiency." J Biol Chem. 275(13):9256-62.*
9. Hirono A et al. (2002). "A single mutation 202G>A in the human glucose-6-phosphate dehydrogenase gene (G6PD) can cause acute hemolysis by itself." Blood. 99(4):1498.*
10. Howes RE et al. (2013). "Spatial distribution of G6PD deficiency variants across malaria-endemic regions." Malar J. 12:418.*

See all references

## Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

| Date | Change |
|---|---|
| **June 4, 2018** | G6PD Deficiency report created. |



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G6PD Deficiency

# Frequently Asked Questions

G6PD deficiency is a common genetic condition caused by defects in an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. The G6PD enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are destroyed upon exposure to certain environmental triggers, which can lead to episodes of anemia. This test includes the most common variant linked to G6PD deficiency in people of African descent.

## G6PD Deficiency

**What does this test do?**

**What does this test not do?**

**The report says the variants included in this test are best studied in people of African descent. What if I'm not of African descent or am only partly of African descent?**

**The report says my genetic result is based on the sex I indicated in my account settings. What does that mean?**

**Where can I learn more about G6PD deficiency, support groups, and other resources?**

**My report says my result could not be determined. What does this mean?**

We could not tell if you have or do not have the tested genetic variant. This can be caused by random test error or other factors that interfere with the test. It can also happen when a person's genetic result is different from what is expected based on the sex they reported in their account settings.

If this result is unexpected, please contact our Customer Care team.

Is this answer helpful? Yes No

**My report says my result could not be determined. What are some things I could do?**

Because we could not determine your result, it is still possible to have the genetic variant tested or another genetic variant not tested.

If this result is unexpected, please contact our Customer Care team.

Consider talking to a healthcare professional if:

- You have a family history of G6PD deficiency or hemolytic anemia.
- You think you might have symptoms of G6PD deficiency.
- You have concerns about your results.

Is this answer helpful? Yes No

**Have more questions?** Check out our Customer Care Help Center.

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